

# WES CRANIOFACIAL ANOMALIES DG 3.4

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
ACP4	227.3	100.0	100.0	617297
ACTG1	270.4	100.0	100.0	No OMIM phenotype
ADAMTSL4	202.1	100.0	100.0	225100
ALX1	184.4	100.0	100.0	613456
ALX3	186.7	100.0	100.0	136760
ALX4	217.6	100.0	100.0	168500
AMBN	176.4	100.0	100.0	616270
AMELX	220.5	100.0	100.0	301200
AMER1	237.8	100.0	100.0	300373
AMTN	171.1	100.0	100.0	617607
ANKRD11	202.6	100.0	100.0	148050
ARHGAP29	160.8	100.0	100.0	-
AXIN2	224.8	100.0	100.0	608615
BCOR	211.7	100.0	100.0	300166
BMP2	196.6	100.0	100.0	-
BMP4	222.6	100.0	100.0	607932
CCBE1	168.7	100.0	100.0	No OMIM phenotype
CDC45	174.4	100.0	100.0	617063
CDON	166.5	100.0	100.0	614226
CDSN	354.1	100.0	100.0	602593
CHD7	185.7	100.0	100.0	214800
COL11A1	149.2	100.0	100.0	154780;604841
COL11A2	274.6	100.0	100.0	215150;277610
COL2A1	179.8	100.0	100.0	108300
COL9A1	162.2	100.0	100.0	614135

COL9A2	190.8	100.0	100.0	614284;600204
COL9A3	204.3	100.0	100.0	120270
COLEC11	215.7	100.0	100.0	No OMIM phenotype
CTSK	177.6	100.0	100.0	265800
CYP26B1	233.7	100.0	100.0	No OMIM phenotype
DHODH	177.3	100.0	100.0	263750
DISP1	191.8	100.0	100.0	-
DLX3	210.3	100.0	100.0	104510
DLX4	221.7	100.0	100.0	616788
DSPP	305.9	100.0	100.0	125490
EDA	197.2	100.0	100.0	305100;313500
EDAR	189.4	100.0	100.0	129490;224900
EDARADD	181.4	100.0	100.0	129490;224900
EDN1	144.4	100.0	100.0	615706
EDNRA	178.0	100.0	100.0	616367
EFNA4	212.6	100.0	100.0	601380
EFNB1	195.6	100.0	100.0	304110
EFTUD2	150.3	100.0	100.0	610536
EIF4A3	161.4	100.0	100.0	268305
ENAM	186.0	100.0	100.0	104500
ERF	227.0	100.0	100.0	600775
ESCO2	142.7	100.0	100.0	268300
EYA1	161.7	100.0	100.0	113650
EZH2	169.0	100.0	100.0	277590
FAM20A	177.5	100.0	100.0	204690
FAM83H	302.7	100.0	100.0	130900
FGD1	196.6	100.0	100.0	305400
FGF10	177.9	100.0	100.0	149730
FGF3	243.5	100.0	100.0	610706
FGF8	200.6	100.0	100.0	612702

FGF9	162.8	100.0	100.0	612961
FGFR1	214.8	100.0	100.0	123150
FGFR2	193.1	100.0	100.0	123500
FGFR3	254.4	100.0	100.0	101400
FLNA	208.6	100.0	100.0	No OMIM phenotype
FLNB	178.2	100.0	100.0	No OMIM phenotype
FOXC1	220.5	100.0	100.0	602482;601631
FOXE1	206.3	100.0	100.0	241850
GDF3	197.0	100.0	100.0	613702
GDF6	189.1	100.0	100.0	118100
GJA1	216.3	100.0	100.0	121014
GJB6	187.9	100.0	100.0	129500
GLI2	235.0	100.0	100.0	610829
GLI3	207.6	100.0	100.0	175700
GNAI3	147.1	100.0	100.0	602483
GNPTAB	172.5	100.0	100.0	No OMIM phenotype
GPR68	247.1	100.0	100.0	617217
GRHL3	164.8	100.0	100.0	606713
GSC	178.3	100.0	100.0	602471
HOXA2	199.4	100.0	100.0	612290
HUWE1	161.8	100.0	100.0	300706
HYAL2	215.6	100.0	100.0	-
IFT122	167.1	100.0	100.0	218330
IFT43	156.0	100.0	100.0	614099
IFT88	134.4	100.0	100.0	-
IKBKG	187.2	100.0	100.0	300291;300301
IL11RA	189.1	100.0	100.0	614188
IL6ST	164.7	100.0	100.0	No OMIM phenotype
IMPAD1	201.7	100.0	100.0	No OMIM phenotype
INTU	150.0	100.0	100.0	617926

IRF6	187.9	100.0	100.0	119300
ITGB6	157.5	100.0	100.0	616221
KAT6B	213.9	100.0	100.0	606170;603736
KDF1	205.8	100.0	100.0	617337
KDM1A	156.6	100.0	100.0	616728
KDM6A	167.6	100.0	100.0	300867
KLK4	188.0	100.0	100.0	204700
KMT2D	227.9	100.0	100.0	147920
KREMEN1	170.0	100.0	100.0	609898
LAMB3	196.0	100.0	100.0	104530
LRP2	170.3	100.0	100.0	222448
LRP6	177.4	100.0	100.0	616724
LTBP3	223.0	100.0	100.0	613097
MASP1	190.9	100.0	100.0	257920
MED12	183.9	100.0	100.0	300895;305450;309520
MEGF8	215.4	100.0	100.0	614976
MEIS2	189.5	100.0	100.0	600987
MEOX1	210.8	100.0	100.0	214300
MID1	181.9	100.0	100.0	300000
MITF	196.6	100.0	100.0	193510
MMP20	154.0	100.0	100.0	612529
MN1	284.5	100.0	100.0	No OMIM phenotype
MSX1	242.7	100.0	100.0	189500
MSX2	213.5	100.0	100.0	168500
NAA10	184.4	100.0	100.0	300013
NECTIN1	208.1	100.0	100.0	225060
NFKBIA	205.2	100.0	100.0	612132
NIPBL	156.1	100.0	100.0	122470
NOG	229.6	100.0	100.0	186500
NSD1	199.8	100.0	100.0	117550

ODAPH	178.1	100.0	100.0	614832
OFD1	130.4	100.0	100.0	311200
OTX2	209.7	100.0	100.0	610125
P4HB	170.6	100.0	100.0	No OMIM phenotype
PAX3	207.8	100.0	100.0	193500
PAX6	190.1	100.0	100.0	602482
PAX7	235.8	100.0	100.0	268220
PAX9	235.7	100.0	100.0	604625
PGM1	157.8	94.2	94.2	614921
PITX2	193.1	100.0	100.0	180500
PLCB4	151.2	100.0	100.0	614669
POLR1C	139.8	83.0	82.8	248390
POLR1D	176.7	100.0	100.0	613717
POR	213.0	100.0	100.0	No OMIM phenotype
PORCN	175.9	100.0	100.0	305600
PRRX1	213.1	100.0	100.0	202650
PTCH1	215.7	100.0	100.0	109400
PTH1R	200.0	100.0	100.0	125350
RAB23	141.4	100.0	100.0	201000
RAD21	167.1	100.0	100.0	614701
RBM10	198.5	100.0	100.0	311900
RECQL4	275.2	100.0	100.0	603780
RIPK4	239.3	100.0	100.0	263650
RUNX2	210.1	100.0	100.0	119600
SALL1	218.3	100.0	100.0	107480
SALL4	209.8	100.0	100.0	607323
SATB2	193.6	100.0	100.0	612313
SCARF2	218.6	100.0	100.0	No OMIM phenotype
SEC24D	163.0	100.0	100.0	No OMIM phenotype
SEMA3E	161.7	100.0	100.0	214800

SF3B2	181.4	100.0	100.0	-
SF3B4	187.0	100.0	100.0	154400
SH3BP2	176.5	99.9	99.4	118400
SHH	230.9	100.0	100.0	147250;611638
SIX1	215.6	100.0	100.0	608389
SIX3	235.5	100.0	100.0	157170
SIX5	227.7	100.0	100.0	No OMIM phenotype
SKI	184.5	100.0	100.0	182212
SLC24A4	165.8	100.0	100.0	615887
SLC26A2	188.6	100.0	100.0	256050
SMAD6	203.7	100.0	100.0	617439
SMC1A	164.8	100.0	100.0	300590
SMC3	142.5	100.0	100.0	610759
SMO	217.9	100.0	100.0	601707
SMOC2	173.0	100.0	100.0	125400
SNAI2	192.8	100.0	100.0	608890
SOX10	254.5	100.0	100.0	613266
SOX6	158.9	100.0	100.0	607257
SOX9	258.7	100.0	100.0	114290
SPECC1L	163.7	97.8	96.2	600251
SUMO1	101.9	69.7	69.4	613705
TBX1	158.0	98.1	95.9	192430
TBX22	201.1	100.0	100.0	303400
TCF12	165.0	100.0	100.0	615314
TCOF1	188.3	100.0	100.0	154500
TFAP2A	192.5	100.0	100.0	113620
TGFBR1	161.3	100.0	99.9	609192
TGFBR2	218.2	100.0	100.0	610168
TGIF1	212.8	100.0	100.0	142946
TLK2	149.1	100.0	100.0	No OMIM phenotype

TP63	184.0	100.0	100.0	604292
TRAF6	174.5	100.0	100.0	602355
TSHZ1	229.8	100.0	100.0	607842
TSPEAR	184.5	100.0	100.0	618180
TWIST1	189.8	100.0	100.0	101400
UBB	273.8	100.0	100.0	119540
VAX1	162.4	99.9	99.5	614402
WDR19	150.1	100.0	100.0	614378
WDR35	151.2	100.0	100.0	613610
WDR72	161.5	96.9	96.9	613211
WNT10A	218.5	100.0	100.0	224750;257980
WNT10B	239.8	100.0	100.0	617073
ZEB2	179.1	97.4	97.4	235730
ZIC1	268.7	100.0	100.0	616602
ZIC2	212.2	100.0	100.0	609637

*Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.*

*TWIST is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.*

*Median Coverage describes the average number of reads seen across 50 exomes.*

*Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.*

*Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.*

*Genes with no value for coverage are non protein coding genes.*

*Non protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.*

*OMIM release used for OMIM disease identifiers and descriptions: September 1st, 2021.*